



Late Diagnosis of Mosaic Turner's Syndrome - Rare Genetic Disease- A Case Report

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ABSTRACT

Turner syndrome is a rare chromosomal disorder affecting 1 in 2,500 females, characterized by complete or partial absence of one X chromosome. Mosaic Turner syndrome, where both normal and abnormal cell lines coexist, often presents with subtle symptoms leading to delayed diagnosis. We present a 22-year-old female with primary amenorrhea, short stature, and absent secondary sexual characteristics. Imaging and karyotyping confirmed mosaic Turner syndrome. She was managed with hormone replacement therapy. This case highlights the diagnostic challenges and emphasizes the importance of early recognition to improve long-term outcomes.

Keywords: Turner's syndrome, mosaic turner syndrome, primary amenorrhea, karyotyping, diagnostic laparoscopy

INTRODUCTION

Turner syndrome (TS) is a chromosomal abnormalities in females, occurring in approximately 1 in 2,500 live female births. It results from complete or partial absence of one X chromosome, leading to a spectrum of clinical manifestations that affect growth, gonadal development, and multiple organ systems (1,2).

Two major cytogenetic variants of Turner Syndrome are recognized:

1. Classical Turner syndrome (45,X): Involves complete absence of one X chromosome, typically associated with characteristic clinical features such as short stature, webbed neck, lymphedema at birth, broad chest with widely spaced nipples, gonadal dysgenesis, and primary amenorrhea.
2. Mosaic Turner syndrome (45,X/46,XX or other variants): Involves a mixture of normal and abnormal cell lines, resulting in variable and often milder phenotypic expressions. Patients may lack the classic dysmorphic features, making diagnosis more challenging and frequently delayed until adolescence or adulthood (3).

The clinical presentation of Turner Syndrome is heterogeneous. While short stature and gonadal failure are the most consistent findings, other features may include cardiovascular malformations (bicuspid aortic valve, coarctation of aorta), renal anomalies, hearing impairment, metabolic disturbances, and autoimmune disorders (4,5). Mosaic forms may present later, sometimes only with infertility or primary amenorrhea, further complicating timely recognition.

Early diagnosis of Turner Syndrome is crucial because it allows timely initiation of growth hormone therapy to improve final adult height, estrogen replacement to induce secondary sexual characteristics, and monitoring for comorbidities. Delayed diagnosis, especially in mosaic cases, often results in missed opportunities for optimal intervention and has significant psychosocial and reproductive implications (6).

Despite increased awareness, many cases of mosaic Turner Syndrome remain underdiagnosed due to subtle phenotypic presentation. In resource-limited settings, lack of early genetic evaluation and late presentation of patients with menstrual complaints further contribute to delayed recognition.



The purpose of this case report is to present a rare instance of Mosaic Turner syndrome diagnosed at 22 years of age, highlight the clinical features that contributed to the delayed diagnosis, and discuss the importance of early recognition and management to improve long-term outcomes.

INCIDENCE

Around 50% of Turner syndrome exhibit monosomy X (45,X), while 5% to 10% possess an isochromosome duplication of the long arm of an X chromosome (46,X,i(Xq)). The majority of the remaining 30% to 40% individuals display mosaicism for 45X/46XX, which is accompanied by one or more alternate cell lineages, which results in unusual clinical presentation.

CASE REPORT

A 22 years old girl resident of Gabbur, Raichur, presented to OBG OPD with Primary Amenorrhoea. She was born out of non-consanguineous marriage. Her paternal cousin had similar complaints. Her siblings are 20 and 26 years old with menarche achieved at the age of 14 years. Her height was 142cm, weight 35 kg with BMI 16.6 kg/m² and apparently a normal intelligence.

General physical examination: showed her vitals were within normal range with female phenotype, short stature, broad chest with wide spaced nipples, low hairline. Her breast examination showed prepubertal development consistent with tanner stage-I, with an absence of axillary hair development also consistent with Tanner's-stage I. (Fig-1) Systemic Examination were unremarkable.

Local examination: showed absent pubic hair suggesting tanner stage-I with infantile hypoplastic labia majora, minora and clitoris (Fig-2). Per speculum examination vaginal orifice normal and cervix normal with pinpoint os.(Fig 3)

Bimanual examination –showed uterus appears knob like structure, smaller than normal size, bilateral fornix is free.

Per rectal examination-normal



Fig-1- short stature, broad chest with wide spaced nipples, low hairline with breast, axillary and pubic hair development of Tanner's-stage I

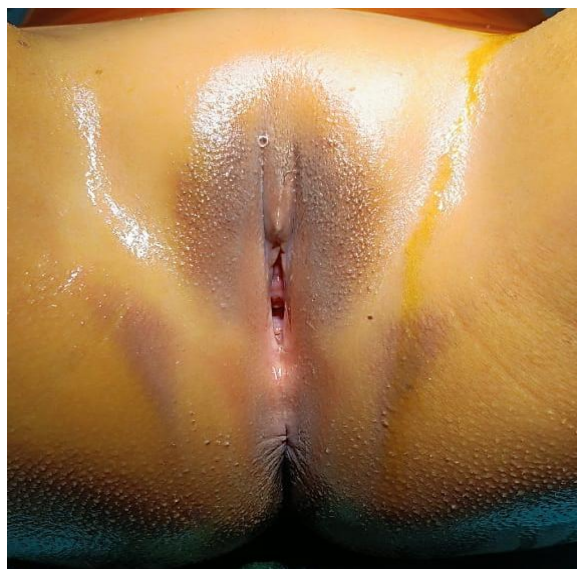


Fig-2-local examination showed absent pubic hair with infantile hypoplastic labia majora, minora and clitoris.

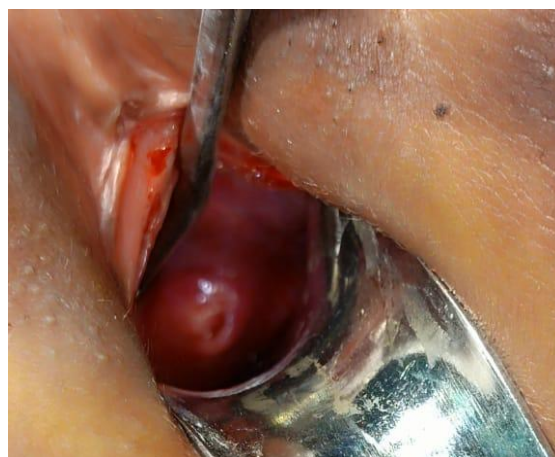


Fig-3-Per speculum examination-vaginal orifice normal and cervix normal with pinpoint os.

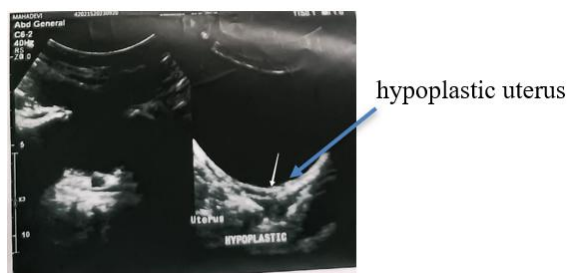
INVESTIGATIONS

Haemoglobin-12.5gm%, Total count- 6,500cells, Blood group and Rh typing- B positive, urine routine – Normal, Coagulation profile- Normal, Serology- Non reactive, RBS- 105mg/dl, TSH-4.62 μ IU/mL, renal and liver function tests-normal, ECG and 2DECHO- was normal.

Hormonal evaluation showed hypergonadotropic hypogonadism with elevated FSH-68.80 mIU/ml and LH-52.71 mIU/ml with low estradiol<10 pg/ml and normal prolactin levels-6.47 ng/ml.

USG Abdomen and pelvis (Fig-4)- showed hypoplastic uterus and ovaries, suggested MRI pelvis for further evaluation.

MRI PELVIS- Hypoplastic uterus measuring 1.7x0.7cms (Fig-5) bilateral streak ovaries right ovary measuring 8.8x4.4mm (Fig-6) and left ovary measuring 8.5x4.6mm(Fig-7) with no evidence of follicles with possibility of turner syndrome.



(Fig-4)USG- hypoplastic uterus

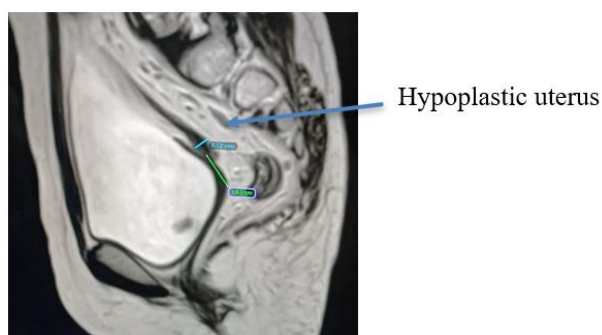


Fig-5-MRI hypoplastic PELVIS- showing hypoplastic uterus measuring 1.7x0.7cms

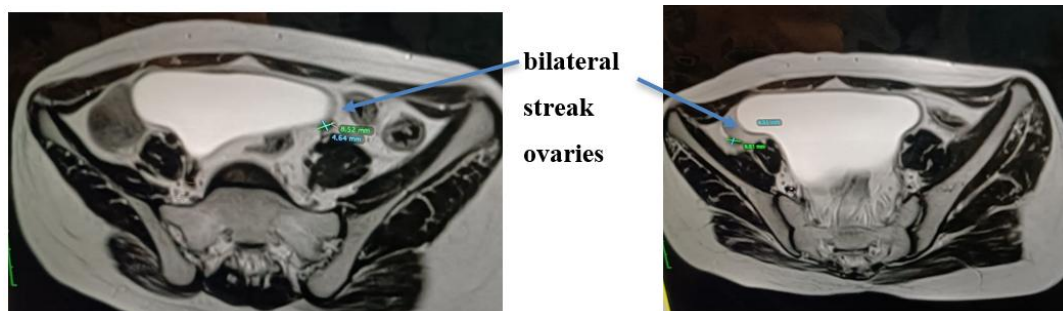


Fig-6 and Fig-7 MRI PELVIS- bilateral streak ovaries right ovary measuring 8.8x4.4mm and left ovary measuring 8.5x4.6mm with no evidence of follicles.

FURTHER EVALUATION THROUGH DIAGNOSTIC LAPAROSCOPY- Small hypoplastic knob like uterus was noted with B/L tubes adherent to pelvic wall. B/L ovaries were not visualized. (Fig-8)

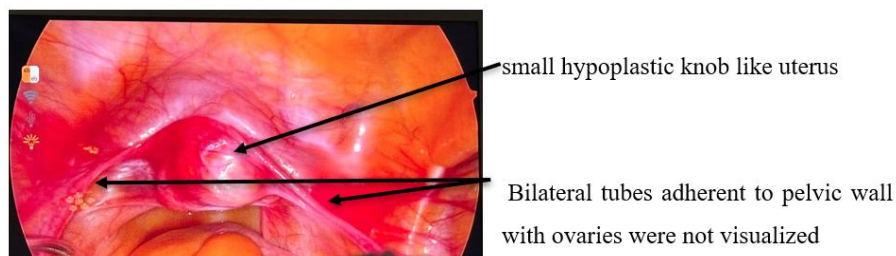


Fig-8-Diagnostic laparoscopy showed-small hypoplastic knob like uterus was noted with bilateral tubes adherent to pelvic wall and bilateral ovaries were not visualized.

KARYOTYPE- Peripheral blood karyotyping revealed a 46XX pattern with mosaicism, supporting a diagnosis of Turner syndrome with mosaicism (Fig-9).

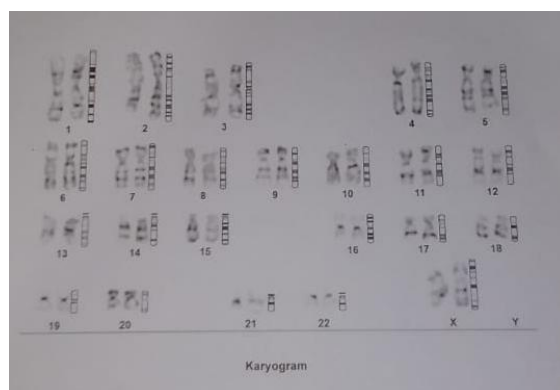


Fig-9-karyotype showed 46XX with mosaicism

DIAGNOSIS- Turner's syndrome with mosaicism

MANAGEMENT-The patient was initiated on hormone replacement therapy with estrogen and advised regular follow-up. Counseling was provided regarding long-term management, fertility implications, and psychological support.

DISCUSSION

Turner syndrome remains a diagnostic challenge because of its heterogeneous presentation, particularly in mosaic cases. Classical Turner syndrome (45,X) is usually identified earlier due to obvious physical features such as webbed neck, broad chest, or neonatal lymphedema. However, mosaic forms, such as 45,X/46,XX, often present later, as their phenotype is milder or atypical (3). In our case, the diagnosis was delayed until 22 years of age, which is uncommon but consistent with literature describing delayed detection in mosaic forms (5,7).

The hallmark features of Turner syndrome include short stature and gonadal dysgenesis, both of which were evident in our patient. Short stature results from haploinsufficiency of the SHOX gene, located on the X chromosome's pseudoautosomal region (8). Gonadal failure in Turner syndrome typically leads to hypergonadotropic hypogonadism, explaining the elevated FSH and LH with low estradiol levels seen in our patient.

Importantly, Turner syndrome is associated with several systemic complications. Cardiovascular anomalies, including bicuspid aortic valve, coarctation of the aorta, and aortic dissection, are found in up to 30–50% of patients and are the leading causes of early mortality (9). Renal malformations, metabolic syndrome, autoimmune thyroid disease, and osteoporosis also occur at increased frequency (10,11). Although our patient's systemic evaluation was normal, long-term follow-up remains critical to monitor for such complications.

Hormone replacement therapy plays a vital role in management, helping induce secondary sexual development, maintain bone health, and reduce cardiovascular risks (6). Ideally, estrogen replacement should begin at the expected age of puberty, but in delayed-diagnosis cases like ours, it is initiated in adulthood. The psychosocial impact of delayed puberty and infertility must not be underestimated, making counseling an essential aspect of care (12).

Several case reports highlight late diagnosis of Turner syndrome, sometimes even in postmenopausal women. Jin et al. described a case diagnosed at 61 years, while Mathuriya and Dave reported a mosaic TS in the third decade of life (7,3). Such reports emphasize the need for heightened clinical suspicion in patients presenting with primary amenorrhea, unexplained short stature, or absent secondary sexual development, even in the absence of classical phenotypic features.

With advances in reproductive technology, women with Mosaic Turner syndrome who have normal uterus may achieve pregnancy using donor oocytes and assisted reproduction. However, this is associated with increased cardiovascular risks and must be approached cautiously, with thorough pre-conception cardiac evaluation (13).

Our case underscores the importance of early diagnosis through careful clinical evaluation, supported by cytogenetic analysis. Greater awareness among clinicians, particularly in resource-limited settings, can help reduce delays, enabling timely initiation of growth hormone and estrogen therapy, and improving long-term health and quality of life.



CONCLUSION

Turner syndrome mosaicism presents with varying symptoms, with severity correlated to the proportion of the affected chromosome, which is typically diagnosed through karyotyping, while laparoscopy is the diagnostic tool to aid in confirming the condition.

Early diagnosis, counselling the patient and timely therapy reduces morbidity and improves outcome. Treatment should be personalized depending upon clinical features, needs of patient and reduce the psychological effect on patient and family.

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